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## Single-nucleotide polymorphisms in the P2x7 receptor gene are associated with bone mineral density and ankle fractures

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### ABSTRACT

**Introduction:** The objective of this study is to determine the associations among genetic variations in the P2X7 receptor gene, decreased bone mineral density (BMD), and the risk of osteoporosis in patients aged older than 50 years with ankle fractures.

**Methods:** Patients were genotyped for 15 nonsynonymous single-nucleotide polymorphisms (SNPs) in the P2X7 gene. The sample was divided into two groups according to the bone densitometry results: an intervention group with osteopenia (T scores between -1.0 and -2.5) or osteoporosis (T scores  $\leq$ -2.5) and a control group with values within the normal range (T scores  $\geq$ -1). A total of 121 patients were evaluated: 65 in the intervention group and 56 in the control group.

**Results:** The results suggested that SNPs 1, 4, 11, 13, 14, and 15 were loss-of-function (LOF) variants. SNP 12 was also associated with LOF in our population, but its RNA expression has not been analyzed to date.

**Conclusions:** In conclusion, we demonstrate that functional polymorphisms in P2X7 are associated with BMD and with an increased risk of ankle fractures. The limitations of our study are its focus on nonsynonymous polymorphisms, which do not cover all genetic variations in P2X7, and its small sample size compared with the international literature. A strength of this study is that it is the first to evaluate P2X7 in the Brazilian population.

**Keywords:** Single-nucleotide polymorphisms; P2X7; Purinergic; Osteoporosis; BMD; Ankle fracture.

